<u>WE CLAIM</u>:

1.   An isolated nucleic acid molecule encoding a protein with a RING-
finger domain and 6 NHL-motifs wherein the protein is associated with
Lafora's disease.

2.   A nucleic acid according to claim 1 having a sequence as shown in
SEQ ID NO:1 (Figure 6A) or SEQ ID NO:3 (Figure 7).

3.   An isolated nucleic acid molecule according to claim 1 comprising

        (a)   a nucleic acid sequence as shown in SEQ ID NO:1 (Figure 6A)
or SEQ ID NO:3 (Figure 7A), wherein T can also be U;

        (b)   a nucleic acid sequence complementary to (a);

        (c)   a nucleic acid sequence that has substantial sequence
homology to a nucleic acid sequence of (a) or (b);

        (d)   a nucleic acid sequence that is an analog of a nucleic acid
sequence of (a), (b) or (c); or

        (e)   a nucleic acid sequence that hybridizes to a nucleic acid
sequence of (a), (b), (c) or (d) under stringent hybridization conditions.

4.   A method of detecting Lafora's disease comprising detecting a
mutation in a nucleic acid sequence according to any one of claims 1 to 3 in a
sample from an animal.

5.   A method according to claim 4 comprising detecting a C to G change at
nucelotide number 205 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

6.   A method according to claim 4 comprising detecting a T to A change at
nucleotide number 76 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

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7.   A method according to claim 4 comprising detecting a deletion of nucleotides GA at nucleotide positions 1048 and 1049 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

5   8.   A method according to claim 4 comprising detecting a deletion of nucleotides AG at nucleotide positions 468 and 469 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

9.   A method according to claim 4 comprising detecting a deletion of 10   nucleotide G at nucleotide number 992 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

10.   A method according to claim 4 comprising detecting a deletion of 10 bp at nucleotide positions 373 to 382 in the *EPM2B* gene sequence shown in 15   SEQ ID NO:1.

11.   A method according to claim 4 comprising detecting a deletion of 32 bp at nucleotide positions 661 to 692 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

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12.   A method according to claim 4 comprising detecting a T to C change at nucleotide number 260 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

13.   A method according to claim 4 comprising detecting a A to C change at 25   nucleotide number 905 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

14.   A method according to claim 4 comprising detecting a T to C change at nucleotide number 98 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

30   15.   A method according to claim 4 comprising detecting an insert of 2 Ts at nucleotide number 892 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

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16. A method according to claim 4 comprising detecting a G to A change at nucleotide number 436 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

17. A method according to claim 4 comprising detecting a deletion of
5 nucleotide T at nucleotide number 1100 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

18. A method according to claim 4 comprising detecting a deletion of nucleotide T at nucleotide position 606 in the *EPM2B* gene sequence shown
10 in SEQ ID NO:1.

19. A method according to claim 4 comprising detecting a A to T change at nucleotide number 923 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

15 20. A method according to claim 4 comprising detecting a G to T change at nucleotide number 580 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

21. A method according to claim 4 comprising detecting a G to T change at nucleotide number 199 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

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22. A method according to claim 4 comprising detecting a G to A change at nucleotide number 838 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

23. A method according to claim 4 comprising detecting a C to T change at
25 nucleotide number 676 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

24. A method according to claim 4 comprising detecting a deletion of nucleotide A at nucleotide position 468 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

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25.   A method according to claim 4 comprising detecting a deletion of nucleotide C at nucleotide position 204 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

5   26.   A method according to claim 4 comprising detecting one or more mutations in the *EPM2B* gene as indicated in Table 1.

27.   A method according to claim 4 comprising detecting a repeat of the sequence GCCGCCCCCGC (SEQ ID NO:5) at nucleotide position 1001 in
10   the canine sequence of *EPM2B* shown in SEQ ID NO:3.

28.   A method according to claim 22 comprising detecting at least 3 repeats of SEQ ID NO:5.

15   29.   A method according to claim 22 comprising detecting at least 10 repeats of SEQ ID NO:5.

30.   A method according to claim 22 comprising detecting from about 14 to about 26 repeats of SEQ ID NO:5.
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31.   A method according to any one of claims 4-26 wherein the animal is human.

32.   A method according to any one of claims 4 and 26-30 wherein the
25   animal is a canid.

33.   A method according to any one of claims 4 and 26-30 wherein the animal is a dog.

30   34.   An isolated protein containing a RING-finger domain and six NHL domains which protein is associated with Lafora's disease.

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35.   A protein according to claim 34 having the amino acid sequence as shown in SEQ ID NO:2 (Figure 6B) or SEQ ID NO:4 (Figure 7).

36.   A method for detecting Lafora's disease comprising detecting a
5   mutation in a protein according to any one of claims 34 or 35.

37.   A method according to claim 36 comprising detecting a mutation in the *EPM2B* protein as indicated in Table 1.

10   38.   A kit for carrying out the method of any one of claims 4 to 33 or 37 comprising reagents for the detection of a mutation in a nucleic acid sequence as shown in SEQ ID NO:1 or SEQ ID NO:3.

39. '   A kit for carrying out the method of any one of claims 36 or 37
15   comprising reagents for the detection of a mutation in a protein sequence as shown in SEQ ID NO:2 or SEQ ID NO:5.